

FOLIA MEDICA CRACOVIENSIA

Vol. LX, 3, 2020: 17–26

PL ISSN 0015-5616

DOI: 10.24425/fmc.2020.135792

## Early fetal cardiac scan as an element of the sonographic first-trimester screening

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**Abstract:** Early fetal cardiac scan (EFCS) is becoming an increasingly common element of the first trimester ultrasound screening carried out at 11–14 gestational weeks. It offers the first possibility to detect congenital heart defects (CHD) or, in ambiguous cases, to identify those pregnancies where a more detailed cardiac scan would be required later in pregnancy. The size of the fetal heart at the end of the first trimester and the associated relatively low image resolution make it impossible to capture all cardiac data to inform the ultimate picture. However, even at this stage, cues of anatomical and functional abnormalities can be picked up, which suggest not only a CHD, but also a likelihood of cardiovascular symptoms typical of genetic disorders. EFCS should focus on cardiac position, atrioventricular (AV) connections, AV valve function, initial assessment of ventriculo-arterial (VA) connections and the presence of red flag signs in the three vessel and trachea view (3VTV). Proper use of color Doppler mapping makes it possible to overcome the low resolution of B-mode to a certain extent. Here we present our long-term experience in EFCS.

**Key words:** first trimester, fetal heart, congenital heart disease, ultrasound screening.

**Submitted:** 22-Sep-2020; **Accepted in the final form:** 30-Oct-2020; **Published:** 30-Nov-2020.

## Introduction

The prevalence of CHDs has remained stable over the recent years in the European Union member states. According to the Central Registry of EUROCAT, the incidence rate of CHD was 80.1 per 10,000 births in 2011, and 78.5 per 10,000 births in 2017 [1]. CHD detection rate is improving with newer educational initiatives, better availability of antenatal screening and including EFCS in the first trimester ultrasound screening protocol [1–5]. EFCS is becoming an increasingly common element of the first trimester ultrasound screening carried out at 11–14 gestational weeks [4]. The first trimester screening is usually associated with risk assessment for major trisomies and pre-eclampsia according to the Fetal Medicine Foundation algorithms. However, the addition of early comprehensive fetal anomaly scan protocol, including EFCS, to routine first trimester screening has become popular in recent years [5–7]. Some authors aimed at identifying indications for EFCS based on markers and anomalies seen at the time of first trimester screening, such as increased nuchal translucency (NT), tricuspid regurgitation (TR) or abnormal ductus venosus flow velocimetry [8–11]. However, these key parameters offered insufficient sensitivity and specificity to be considered effective screening for CHD. On the other hand, there is published evidence to support improved detection of congenital defects, including CHD, and improved efficacy of screening for genetic disorders where EFCS has been a part of the first trimester screening [12–14].

Therefore, clinicians carrying out the first trimester screening including EFCS should be aware of both its advantages and limitations. Despite full structural development, fetal heart is very small at this gestational stage and, therefore, B-mode sonography may not yield satisfactory effects. Hence, a clinician needs to acknowledge a significantly lower image resolution than in scans later in pregnancy. For that reason, some authors recommend flow mapping using color Doppler and bidirectional power Doppler as key aspects to complement the B-mode scan. It should be noted that flow mapping methods need to be used during the first trimester screening in line with safety recommendations, such as monitoring the Thermal Index (TI) and the Mechanical Index (MI), as well as reducing the assessment duration to the absolute minimum [15, 16]. It is a significant difference compared to fetal cardiac imaging later in pregnancy, where flow mapping obviously complements the B-mode, but it is less detailed. Based on the available literature, we support this approach to EFCS and recommend starting the scan by placing the probe against the fetal chest, so that the fetal spine is located at the 6 or 12 o'clock position and the ultrasound beam is directed at the interventricular septum at about 45 or 135 degrees. This ensures a proper angle for mapping the inflow in the four-chamber view (4CV) and a transverse view of ductal and aortic arches in 3VTV. As a result, it is possible to image the key fetal geometric cardiovascular determinants, that is, the system of atria and ven-

tricles, as well as aortic and ductal arches, which form the V-sign in 3VTV. Between the two abovementioned views, there is the three-vessel view (3VV) which in normal anatomical conditions shows that the cross-section through the arterial duct originates from the ventricle located anteriorly and through ascending aorta (Fig. 1).



**Fig. 1.** Basic views commonly used in EFCS with color Doppler. From the left: four-chamber view, three-vessel view, and three-vessel and trachea view.

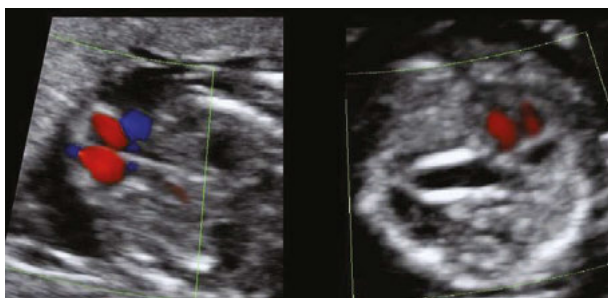
From the practical perspective, if color Doppler signal is attenuated or an attempt is made to confirm its presence, a translation maneuver should be used so that the structure in question is seen as vertical on the screen to optimize the angle and improve the sensitivity of color Doppler flow mapping. Due to significantly lower image resolution, clinicians need to be aware that some diagnostic questions and queries may remain unanswered during the first trimester screening. Nevertheless, it is the first moment when CHD can be either detected or suspected. In the latter case, mid-trimester cardiac scan should be aimed at verifying the diagnosis. In the current article, we share our long-term experience of EFCS, discussing its practical aspects and limitations [9, 10, 12, 17].

### Assessment protocol

Considering the possibilities of EFCS, the clinician should seek answers to the following diagnostic questions:

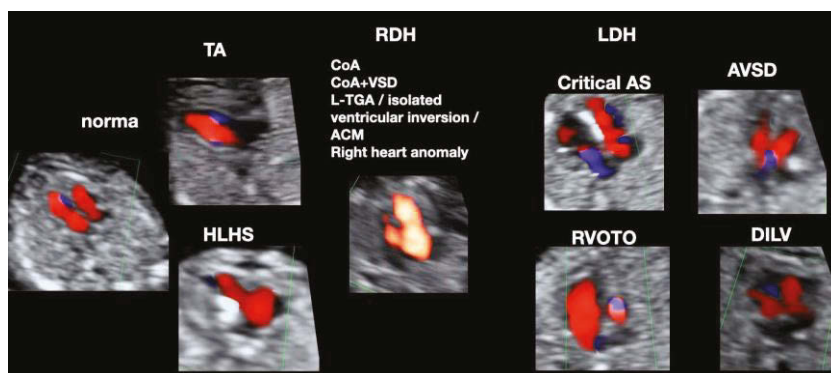
1. Is the situs normal, that is, is the majority of the heart located on the left side of the chest?
2. Do the atrioventricular connections appear normal or abnormal?
3. What is the function of atrioventricular valves like?
4. What is the preliminary image of the ventriculo-arterial connections based on their geometry?
5. Are there any red flag signs present in the three vessel and trachea (3VTV)?

Considering the above clinical questions, the clinician should initially exclude any abnormal heart position e.g. due to an extensive left-sided diaphragmatic hernia causing dextrocardia or pentalogy of Cantrell with an extensive abdominal wall defect, lower sternal defect and ectopia cordis (Fig. 2).



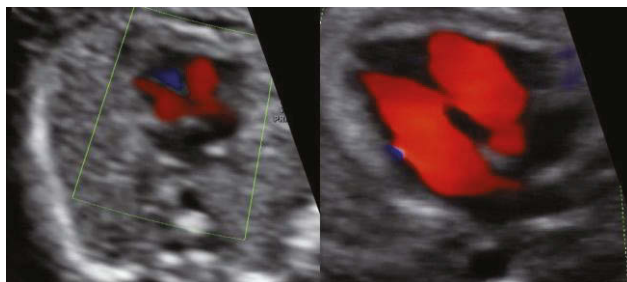
**Fig. 2.** Abnormal cardiac position seen in EFCS. Left: ectopia cordis. Right: large, left-sided diaphragmatic hernia with cardiac dextroposition.

The next step is to exclude situs ambiguous or total situs inversus. Furthermore, the ventricular filling pattern(s) need to be determined, which are crucial for assessing the atrioventricular valve connection (Fig. 3).



**Fig. 3.** The most common patterns of atrio-ventricular connections seen in EFCS. Abbreviations: HLHS — hypoplastic left heart syndrome; TA — tricuspid atresia; RDH — right dominant heart; CoA — coarctation of the aorta; VSD — ventricular septal defect; L-TGA — levo-transposition of the great arteries; ACM — anatomically corrected malposition of the great arteries; LDH — left dominant heart; RVOTO — right ventricle outflow tract obstruction; AVSD — atrioventricular septal defect; AS — aortic stenosis; DILV — double inlet left ventricle.

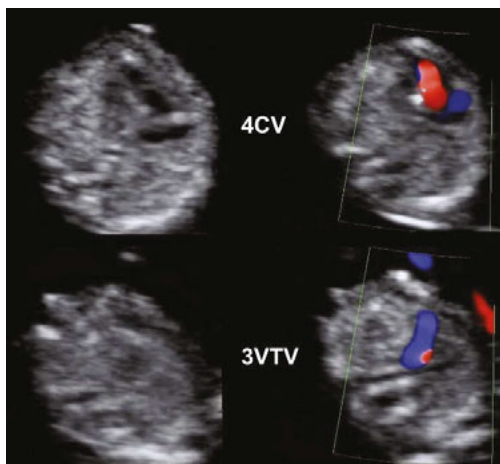
The normal fetal ventricular filling pattern seen in EFCS involves each ventricle receiving blood from a separate atrium and the inflow to the ventricle located more anteriorly being shorter. The width of the inflow area should be comparable between the ventricles. It should be noted that in double inlet ventricle (DIV), two antegrade flows filling a single ventricle are visualized. When color Doppler-mapped flow is not correlated with the background B-mode, the image may be misinterpreted (Fig. 4).



**Fig. 4.** Double inlet left ventricle (DILV) seen in color Doppler during first trimester screening (left) and mid-trimester screening (right).

The right dominant heart (RDH) pattern also requires further investigation. This is most commonly caused by aortic arch obstruction, with or without a ventricular septal defect (VSD), which is formally diagnosed as either coarctation of the aorta, aortic arch hypoplasia or interrupted aortic arch later in pregnancy. So, we recommend 3VTV as the first one to assess for a dominance of the ductal arch over the aortic one, a finding which may support the preliminary suspicion of aortic arch obstruction. It should be noted that the right dominant pattern may stem from atrioventricular discordance termed also as ventricular inversion seen in: the levo-transposition of the great arteries (L-TGA); isolated ventricular inversion or some forms of anatomically corrected malposition (ACM) of the great arteries. The right dominant pattern seen in the first trimester screening may also be associated with double-outlet right ventricle (DORV). However, the defect cannot be comprehensively assessed at this gestational stage. Furthermore, right heart defects, especially tricuspid valve abnormalities, such as Ebstein anomaly (EA) or tricuspid valve dysplasia (TVD), may also result in right heart dominance seen in ultrasound in the first trimester imaging. In this group of defects, a significant tricuspid regurgitation is seen during systole.

On the other hand, if left heart dominance pattern is seen with peripheral vascularity greater than central in flow mapping with color Doppler or power Doppler, it is a feature of critical aortic stenosis. Comorbid mitral regurgitation can also be seen in majority of cases. The details of the ventricular outflow tract are not always easily assessed with color Doppler or spectral Doppler in EFCS during the first-trimester ultrasound screening. If univentricular AV connection is suspected, the possible diagnoses are hypoplastic left heart syndrome (HLHS) and tricuspid atresia (TA). Where a single inflow is visualized with an irregular flow contour periapically, 3VTV should be used for a thorough assessment. A dilated vertical pulmonary trunk and arterial duct seen in 3VTV are suggestive of hypoplastic left heart syndrome (HLHS). Lowering the color Doppler baseline enables visualizing the retrograde blood flow in the hypoplastic aortic arch. Possible findings are shown in Fig. 5.



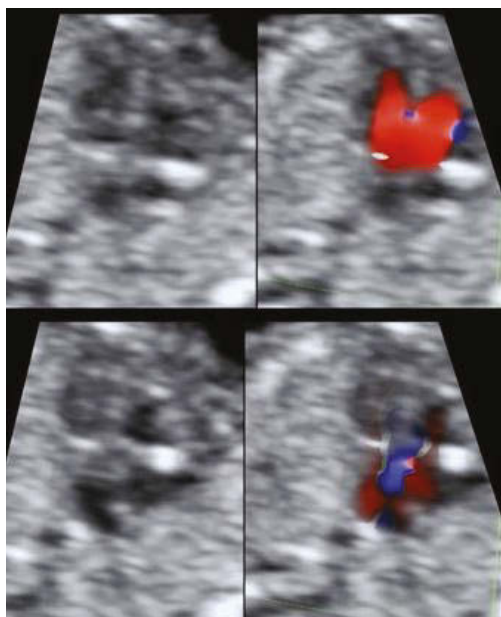
**Fig. 5.** Hypoplastic left heart syndrome (HLHS) seen during EFCS in a B-mode with color Doppler mapping. Abbreviations: 4CV — four-chamber view; 3VTV — three-vessel and trachea view.

On the other hand, a univentricular AV connection with a regular flow contour periapically is primarily suggestive of tricuspid atresia. Unless the ventricular septal defect is large, it will be difficult to visualize in EFCS. In cases of TA, different variants of VA alignments are possible: concordant, dextro- or levo-malposition. The most common variant presents concordant ventriculo-arterial connections with pulmonary stenosis.

A complete atrioventricular septal defect (AVSD) with a significant muscular component may mimic a univentricular AC connection in the first trimester screening. What differs them, though, is the image of the common atrioventricular valve and a large atrioventricular septal defect seen centrally in a B-mode, as well as a ventricular inflow pattern, initially common, which bifurcates slightly laterally together with a medial regurgitant jet seen during systole (Fig. 6).

Late in the first trimester, trivial TR can often be seen. However, more significant cases should alert the clinician to the risk of serious CHD esp. if it coincides with NT thickening (Fig. 7).

TR is detected in the first trimester in about 7% of fetuses [18]. However, in most cases, it is clinically non-significant and resolves spontaneously. Significant TR, where regurgitant jet area exceeds 50% of the atrial surface area, may have several causes, such as right heart defects, conotruncal defects, infection, anemia or extracardiac congenital abnormalities. TR coexistent with increased NT is the most worrying, as it may not only indicate CHD but also chromosomal aberrations. If seen in EFCS, mitral regurgitation should warrant particular caution, just as it does in mid-trimester or third-trimester screening, because it may be a good indicator of left ventricular outflow tract obstruction (LVOTO), mainly at the valvular level. The regurgitation of both atrioventricular valves found in the first trimester fetal cardiovascular imaging is



**Fig. 6.** Atrioventricular septal defect (AVSD) seen in a B-mode with and without color Doppler mapping during diastole (upper) and systole (lower).

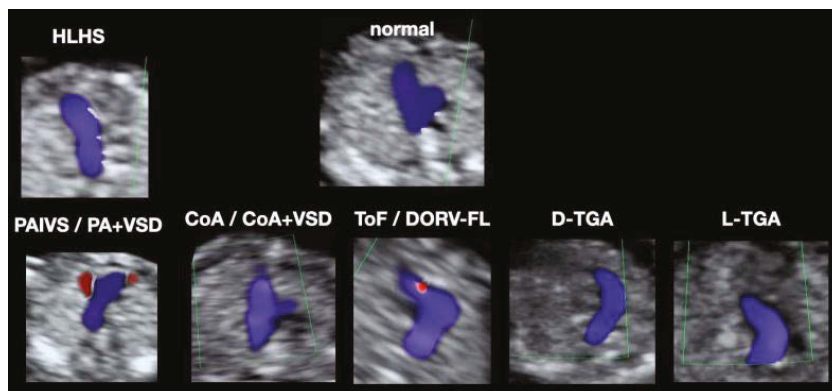


**Fig. 7.** Significant atrioventricular valve regurgitation in EFCS. Left — significant tricuspid regurgitation; Center — mitral regurgitation; Right — common valve regurgitation in atrio-ventricular septal defect.

pathognomonic for a serious fetal condition and usually concomitant with fetal hydrops. In EFCS, just as later in pregnancy, a central regurgitation of common atrio-ventricular valve is typically seen. It is also one of the common features of AVSD seen in the first trimester.

The analysis of 3VTV with color Doppler flow mapping is a key element in EFCS. It ascertains the presence of the V-sign consisting of ductal and aortic arches, assesses their size ratio for potential prominence and the direction of flow in each arch. The most common variants are shown in Fig. 8.





**Fig. 8.** The most common ultrasound findings in the three vessel and trachea (3VTV) with color Doppler flow mapping in EFCS. Abbreviations: HLHS — hypoplastic left heart syndrome; CoA — coarctation of the aorta; VSD — ventricular septal defect; ToF — tetralogy of Fallot; DORV-FL — Fallot-like double outlet right ventricle (= double outlet right ventricle with subaortic ventricular septal defect, subpulmonary infundibulum, and pulmonary stenosis); D-TGA — Dextro-transposition of the great arteries; L-TGA — Levo-transposition of the great arteries; PAIVS — pulmonary atresia with intact interventricular septum; PA+VSD — pulmonary atresia with interventricular septal defect.

A significant prominence of the ductal arch over the aortic arch may suggest coarctation of the aorta or hypoplastic aortic arch, especially if seen in conjunction with right dominant heart in 4CV. The opposite situation, where the aortic arch is more prominent than the ductal arch, especially where ventricular filling appears normal in 4CV, can suggest tetralogy of Fallot (ToF). The absence of the V-sign at the expected location of ductal and aortic arches connection in the 3VTV acquired under optimum conditions is one of the most concerning findings of EFCS. If only one arm of the V sign is seen, the course of the identified arterial vessel needs to be mapped in relation to the symmetrical transverse section through the fetal thorax. For instance, a vertical course of a single arterial trunk in the 3VTV may indicate HLHS, especially if concomitant with single-inlet ventricle. However, if a right-sided convexity of the single arterial trunk is seen in the 3VTV, then D-transposition of the great arteries (D-TGA) should be considered. Finally, a horizontal course of a single arterial trunk may suggest common arterial trunk (CAT). It should also be noted that the aortic or ductal arches with bidirectional flow seen in the first trimester screening constitutes a red flag sign for the risk of fetal demise.

## Conclusion

EFCS should be considered an additional element of antenatal screening for CHD. The possibility to include it, alongside screening for trisomies and pre-eclampsia, in a standard first trimester screening protocol supports its routine use. Fetal cardiac



scan at this gestational stage is a screening procedure aimed at detecting different subtypes of univentricular heart, conotruncal anomalies or CHDs evolving in utero. It significantly improves early detection of chromosomal aberrations [19–21]. Due to its limitations, EFCS cannot replace the mid-trimester fetal echocardiography, which still remains a standard in contemporary antenatal care.

### Conflict of interest

None declared.

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